

Genetic Perspective on Language Replacement in Siberia

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1 Introduction

The sharing of language, cultural traditions and/or religion and legends of common ancestors remain as key elements in identifying modern human ethnic groups (Anderson 2006). But as indicated by history, most modern ethnic groups are admixtures of several ancient populations over different historical periods. In recent decades, the Y-chromosome has proven to be a powerful tool in tracing the paternal history of human ethnic groups and genealogical ancestors. The most common scenario is that the distribution of human languages correlates well with Y-chromosome types (indicating male lineages), whereas mtDNA types (associated with maternal descent) don't correlate with languages (Forster and Renfrew 2011). While most ethnic groups from a language branch tend to share the same predominant paternal lineages, there are also exceptions in some cases worldwide.

There are few historical records on the demographic history and languages of the populations living in Siberia. Hence, it is hard to reveal the detailed ethnic history of these populations and the founding process of their languages. A genetic approach will provide a new perspective for such studies. As the language replacement under the elite dominant mode usually took place in very structured

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D. Xu and H. Li (eds.), *Languages and Genes in Northwestern China and Adjacent Regions*, DOI 10.1007/978-981-10-4169-3_3

populations with a social hierarchy during the historical period (Renfrew 1998), language replacement events or population admixture has left clear genetic mixed signals in the gene pool of present day ethnic groups. We can use genetic signals to assess whether ancient populations interacted. In this study, we have summarized all available Y-SNP haplogroup frequencies for Siberian populations from published research and explored the possible language replacement events in this region. Furthermore, we also briefly discussed the founding process of the Yeniseian language Family, the Altaic language Family (including Turkic, Mongolic, and Tungusic branches), and some other languages.

2 Criteria for Judging Language Replacement

Language replacement, also known as language shift, is a cultural phenomenon in which a human community changes their predominant language to another language, which can be caused by different mechanisms. One of the most common mechanisms is the elite dominant language replacement where a human community gives up their own language and adopts a language with “higher status” in their social hierarchy system (Renfrew 1998). On the other hand, a language replacement can also occur when immigrants reside in a new location that is surrounded by communities speaking another dominant language (hereafter called “integration mode”) (Ehret 1988).

In this research, we generally use two policies to demonstrate a language replacement event:

- A. For most ethnic groups from a language branch, there is one predominant paternal lineage that had undergone a recent expansion. On the contrary, other ethnic groups from the same language branch have different predominant paternal lineages.
- B. For most ethnic groups from a language branch, there are **two or more** predominant paternal lineages that have undergone recent expansions at generally the same historical period. On the contrary, other ethnic groups from the same language branch have different predominant paternal lineages. For this situation, we separate the language replacement into two stages. One of the stages is that language replacement happened before the formation of the common ancestor group of a language branch. In a later period, this language branch spread into a vast geographic region, participating with parallel demic expansions of several predominant paternal lineages. The other stage can be judged by the criterion A.

The comparison of Y-chromosome haplogroup frequencies and the accurate expansion age of paternal lineages in ethnic groups are two key factors in making the judgment of language replacement. In this research, we will also combine achievements from other sub-disciplines of anthropology.

3 Summary of Paternal Genetic Data

To obtain a comprehensive picture of the paternal gene pool of Siberian populations, Y-chromosome haplogroup frequencies of 131 populations living in Siberia or nearby regions were collected from the literature (Chen et al. 2011; Derenko et al. 2006a; b, 2007; Hammer et al. 2006; Hong et al. 2006; Karafet et al. 2001, 2002; Katoh et al. 2005; Khar'kov et al. 2008; Lell et al. 2002; Malyarchuk et al. 2010, 2013; Mirabal et al. 2009; Nasidze et al. 2005; Pakendorf et al. 2006; Pimenoff et al. 2008; Puzyrev et al. 2003; Rootsi et al. 2007; Shi et al. 2008; Shou et al. 2010; Su et al. 1999; Tajima et al. 2004; Tambets et al. 2004; Wells et al. 2001; Xue et al. 2006; Zerjal et al. 2002; Zhong et al. 2010, 2011; Zhou et al. 2008) (Table 1). Since the tested Y-SNP markers varied in different research, we aligned the frequencies of Y-chromosome haplogroups according to the phylogenetic tree on <http://www.isogg.org>. Based on the comparison of Y-chromosome haplogroup frequencies and the expansion age of each paternal lineage, we will discuss possible language replacement events in Siberia in the following sections.

4 Possible Yeniseian Origin of Selkup

The Selkup language belongs to the Samoyed branch of the Uralic language Family (Lewis et al. 2015). The Selkups live by the Taz River and between the middle reaches of the Ob and the Yenisey in Siberia (Kolga et al. 2013). According to previous research, Y-chromosome lineages N1b-P43 and N1c-M178 are two predominant paternal lineages of Uralic-speaking populations (Rootsi et al. 2007), as shown in Table 1. The highest frequencies of haplogroup N1b-P43 were observed in Samoyed populations, like Ngansan (91.1%) and Nenets (74.6%). This haplogroup is also the dominant paternal lineage of Khanty and Mansi populations. On the contrary, Selkups have minor percent of N1b-P43 (6.9%), while haplogroup Q-M242 (66.4%) is the most frequent paternal lineage in this population. Moreover, haplogroup Q is the predominant lineage of the Kets (93.8%), the only living group of the Yeniseian language family. Haplogroup Q-M242 and haplogroup N1b/N1c split around 50,000 years ago. These two haplogroups represent different demographic histories in different regions of the Eurasian continent.

Genetic data shows that the Q-M242 clade in Selkups is one branch of Q1a3a3-L330 found in the Kets population. As seen in Fig. 1, the total age of haplogroup Q1a3a3-B287 is about 5000 years, while the Selkup sample split from the Kets sample at a relatively late age (about 2700 years ago). Given the fact that the Yeniseian language once spread widely along the Yenisei River and the Selkup's range is close to the Yenisei River, it's highly likely that the Selkup population originated from a Yeniseian-speaking group. After their migration into the settlement region of Samoyed populations, they changed their language at an unknown historical period.

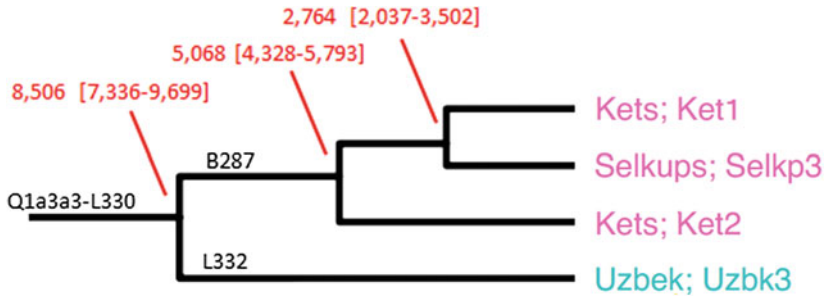


Fig. 1 Phylogenetic tree of paternal lineage Q1a3a3-L330 in Kets and Selkups, based on full Y-chromosome sequences. Number in red indicates the splitting time of downstream branches. Modified from figure S3 of Karmin et al. (2015)

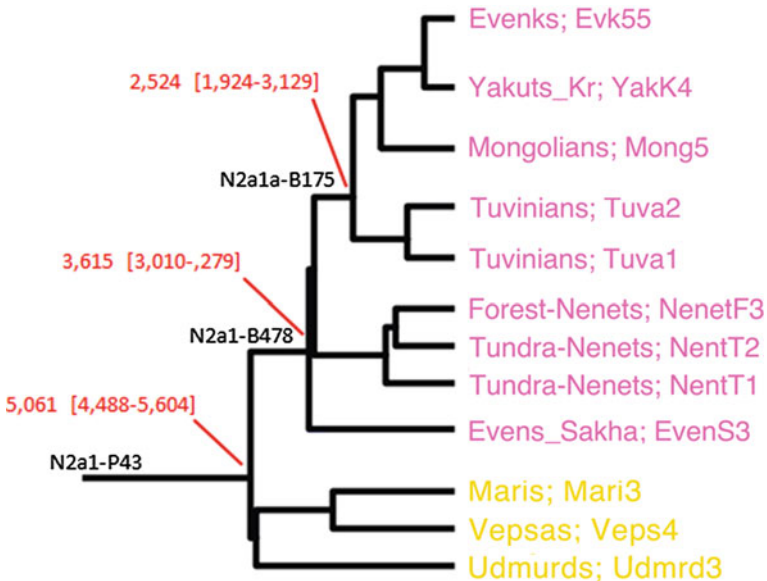


Fig. 2 Phylogenetic tree of N1b-P43 in Samoyed, Tuvan, and other populations, based on full Y-chromosome sequences. Numbers in red indicate the splitting time of downstream branches. Modified from figure S3 of Karmin et al. (2015)

5 Khakassian and Shors

There are four predominant paternal lineages in these two populations, including N1b-P43, N1c-M178, Q-M242, and R1a1a-M17. According to previous research, N1c-M178 and Q-M242 may have lived in south Siberia since the Paleolithic age (Dulik et al. 2012; Rootsi et al. 2007). In addition, ancient DNA shows that haplogroup R1a1a-M17 has been the predominant lineage in the Altai Mountain region

since the Andronovo Culture period (Keyser et al. 2009). Nowadays, there is no Samoyed-speaking population in the Minusinsk Basin and Sayan mountain region. But some ethnology books record that there were some Samoyed tribes classified as Southern Samoyed in the Minusinsk Basin and Tuva until the 17 century (Hajdú 1968; Potapov 1969). They became extinct during later periods. From a genetic perspective, N1b-P43 samples in Samoyed and Tuvan populations belong to a specific sub-clade named N2a1-B478. The expansion time of N2a1-B478 is only about 3600 years ago, as shown in Fig. 2. Hence, we propose that the southern part of Samoyed populations may have changed their language to a Turkic language at various historical periods, bringing haplogroup N2a1-B478 into Tuvan, Khakassian, and Shors populations.

6 The Tuvans and Tofalars

The Tofalars live in the northern region of the East Sayan Mountains. They are assumed to be close relatives of Tuvans. Also, some research lists Tadjins and Sojots as separate ethnic groups distinct from the Tuvans. But usually these two groups are considered as clans of the Tuvans (Vainshtein 1980). According to the 16 sets of data in Table 1, the frequencies of the Y-SNP haplogroup varied a lot in Tuvans samples from different research. Haplogroups N1b-P43, Q-M242, R1a1a-M17, and N1c-M46 can be considered as predominant paternal lineages in Tuvan populations. Additionally, lineage C3-M217 (or C-M130) is quite frequent in Tuvan populations. The Tofalar paternal gene pool is similar to that of the Tuvans. On the contrary, Tuvan-Sojots was found to have a high frequency of C3d-M407 in reference 17.

The high diversities of paternal lineages among the Tuvans and Tofalars are consistent with the complex demographic history of South Siberia, especially the Sayan Mountain and Altaic Mountain region. Previous research showed that Paleolithic populations in South Siberia may be ancestors of most modern ethnic groups in North Eurasia and of Native Americans (Raghavan et al. 2014). Many ancient populations once lived in this region, including populations of the Afanasievo Culture, Andronovo Culture, Chermurchek Culture, and a series of succeeding cultures. In the recent historical period, various populations have lived in this region, including Yeniseian, Samoyed, Turkic, and Mongolic populations. All these populations should have left genetic signals among the Tuvans, Tofalars, and their relatives. Therefore, language replacement may have occurred many times during the historical period in this region.

As discussed in the section on the Khakassian and Shors, some of ancient southern Samoyed populations may have changed their language to the Tuvan language and became part of Tuvan populations. More Y-chromosome sequencing data are needed to reveal a clearer scenario of demographic and linguistic transition in this region since ancient times. Since the C3d-M407 haplotype of the Tuvan-Sojots belongs to the Buryats special lineage and the genetic diversity of

C3d-M407 in Tuvan-Sojots is extremely low, we can assume that a Mongolic-speaking population group (possible Buryats) may have changed their language to a Turkic language when they moved westward into the Sayan Mountain region and finally became a part of Tuvan populations.

7 Kyrgyz and Kazak

The predominant paternal lineage of the Kyrgyz is R1a1a-M17, as seen in Table 1. Karmin et al. 2015 showed that R1a1a-M17 samples in Kyrgyz are classified into two sub-clades (Karmin et al. 2015). One is R1a2a2-B116, a brother branch of R1a2a2-B115 in Altain populations. The other lineage is R1a2c'd-Z2125 which is distributed widely in populations from Central Asia, South Asia and the Middle East. Additional unpublished data from our lab shows that the N1b-P43 and C3*-Star clusters are also important Kyrgyz lineages. We propose that R1a2a2-B116 and N1b-P43 represent the connection of Kyrgyz with populations in South Siberia. On the contrary, R1a2c'd-Z2125 and C3*-Star cluster represent admixture after their migration into the region where they currently reside. More importantly, the Fuyu Kyrgyz language, found in Fuyun County in Heilongjiang province of China, is close to the Khakas language of South Siberia. Since historical records show that present-day Kyrgyz in Kirgizstan originated from the South Siberian region, we propose that the original language of Kyrgyz in Kirgizstan was close to some Turkic languages in South Siberia (perhaps from the “Northern cluster” of the Turkic language group). After their migration to the region where they currently reside, their language changed and became close to Kazak, which belongs to the “Western cluster” of the Turkic language group.

For Kazak populations, a high frequency of haplogroup C3*-M217 can be observed in nearly all research, as seen in Table 1. Further studies showed that most C3*-M217 samples in Kazaks belong to the C3-Star cluster (Abilev et al. 2012). The distribution of this lineage in Eurasia had been proposed to be associated with activities of Chengisz Khan or his close relatives (Zerjal et al. 2002). As summarized in unpublished research at our lab, C3-Star cluster samples in Kazaks are related to Mongolic tribes in the Eastern Chagatai Khanate (A.D. 1348–1514). During the founding process of the Kazak Khanate, a number of Mongol tribes in Eastern Chagatai Khanate moved to the west and joined the Kazak Khanate alliance where a Turkic language (the Kazak language) is the official one. More recently, they have become important parts of modern Kazak populations. Nowadays, the tribal structure is still very strong among Kazaks. So, we propose that the high frequency of the C3-Star cluster was brought in a language replacement event by Mongolic-speaking tribes during the founding process of the modern Kazak ethnicity. Admixture of language elements may also have occurred during the integration of different ethnic groups. From a genetic perspective, it's unreasonable to trace the common words from the Turkic language group and the Mongolic language group in the modern Kazak language.

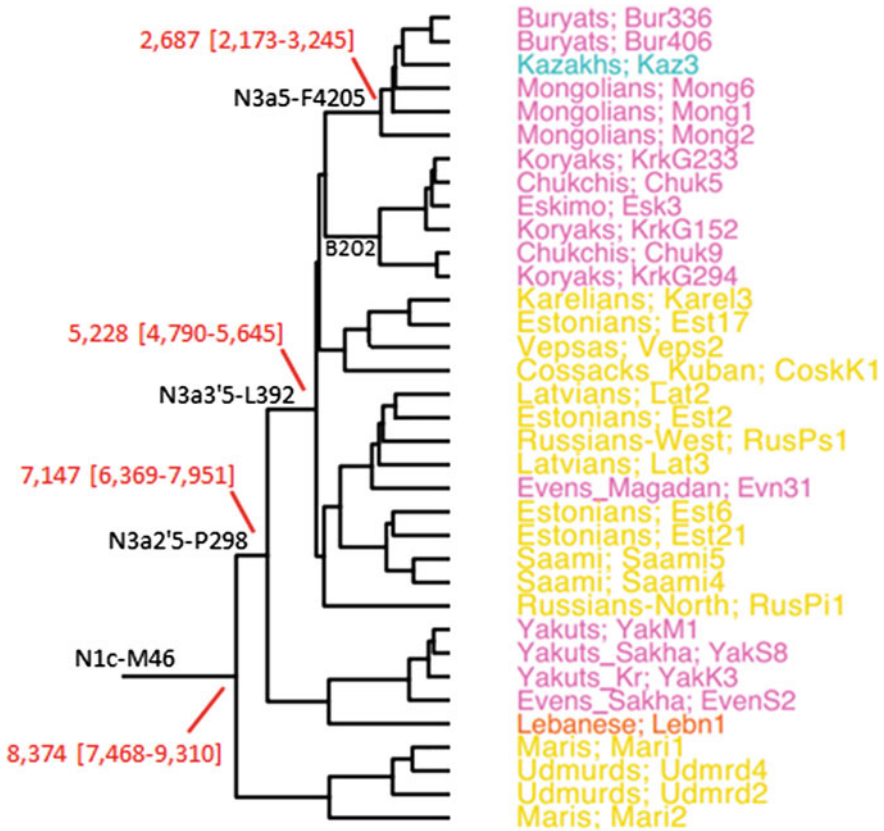


Fig. 3 Phylogenetic tree of paternal lineage N1c-M46 in North Eurasian populations, based on full Y-chromosome sequences. Numbers in red indicate the splitting time of downstream branches. Modified from figure S3 of Karmin et al. (2015)

8 Yakuts

As seen in Table 1, N1c-M46 is the predominant paternal lineage in Yakut populations. Samples of N1c-M46 among Yakuts belong to a special sub-clade named N3a2'5-P298 (Karmin et al. 2015). This sub-clade split from its brother branch N3a1-B211 about 7000 years ago, as shown in Fig. 3. On the contrary, most N1c-M46 samples in Turkic and Mongolic populations belong to another lineage (N3a1-B211). According to archaeological studies, remains of an ancient seminomadic population from the Lake Baikal region, identified as the Turkic-speaking Kurykan people, exhibit close affinities to Yakut culture (Konstantinov 1975; Okladnikov 1970). Hence, it's generally accepted that Yakut populations are descendants of Turkic-speaking populations in the south of their present-day living region. However, N1c-M46 only comprises a minor part or zero present of paternal

lineages in other Turkic populations. As proposed by linguists, diffusion of the Turkic language was largely a result of an “elite dominant mode” during the Turkic Khanate period (A.D. 552–744). Although the paternal lineages of the ruling family of the Turkic Khanate (Ashina clan) is still unknown, we propose that ancestors of Yakut may have changed their language from an unknown one to a Turkic language at an unknown historical period.

9 Mongolian, Kalmyk, Buryat and Khamnigan

As seen in Table 1, Mongolian, Kalmyk, Buryat, and Khamnigan populations have three predominant paternal lineages: C3*-M217, C3c-M48, and C3d-M407. Deep research indicates that most C3*-M217 samples in Mongolic-speaking populations belong to C3-Star cluster which was considered to be the paternal lineage of Chengis Khan or his relatives (Malyarchuk et al. 2013; Zerjal et al. 2002). Hence, we propose that this clade is the core paternal lineage of Mongolic populations. On the other hand, other lineages including D-M174, O-M175, N1b-P43, N1c-M46, Q-M242 and R-M207 also comprise considerable frequencies in the paternal gene pools of these Mongolic-speaking populations. Since many ancient populations once lived on the Mongolian Plateau and Eurasian steppe, these haplogroups may represent the demographic background before the expansion of Mongolian. Also, we cannot rule out the possibility of recent admixture during the historical period from other populations into Mongolic-speaking populations.

For Buryat populations, the high frequency of N1c-M46 can be observed in samples from all research. According to their own history, the original Buryat alliance consisted of the Khor-Tumed tribe, the Bargu tribe and some other hunter-gatherer clans (Abide 1982). The ancient “Bargu” tribe was considered to be a descendant of the Bayarqu (or Bayegu/Baiyrku), a Turkic-speaking tribe in the Turkic Khanate (Tsydendambaev 1972). On the other hand, “Tumed” is possibly an ancient Mongolic word. So, we propose that a Turkic-speaking population (a.k. Bayarqu->Bargu) changed their language and finally became an important part of modern Buryat populations.

The history of the Khamnigan (or Hamnigan), a Mongolic-speaking population living near Nerchinsk, remains unambiguous. Ethnologists believe that the Khamnigans originated from the Tungusic speaking Evenks tribe and underwent Mongolization in the early 16th century (Shubin 1973). But according to genetic data, Khamnigans have a high frequency of C3d-M407 (52.9%) and they do not have haplogroup C3c-M48 which is the predominant lineage of Tungusic speaking populations. We can also predict that the Khamnigan should have a high frequency of N1c-M46 (33.3%), given the Y-chromosome haplotype of this population (Malyarchuk et al. 2013). The two predominant lineages in Khamnigan, C3d-M407 and N1c-M46, make them very close to the Buryat population. Hence, we propose that the Khamnigan were probably originally a Mongolic-speaking population (close to ancestor of Buryats). It may be unreasonable to argue the Tungusic origin

of this population. But on the other hand, we cannot rule out the possibility that they changed to a Tungusic language at an unknown historical period and underwent a re-Mongolization process in the early 16th century.

10 Evenks and Evens

Evenks and Evens were indigenous populations in Siberia before the expansion of Yakuts, living in a vast region ranging from Kamchatka Peninsula in the East to Tunguska River region in the West (Naumov 2006). All available data indicate that haplogroup C3c-M48 is the predominant paternal lineage of Evenks and Evens, as well as other Tungusic-speaking populations. But Yakut-speaking Evenks from Pakendorf et al. 2006 have high frequency of N1c-M46 (72.7%) and low frequency of C3c-M48 (12.1%) (Pakendorf et al. 2006). This population traces their ancestors to the Yakuts, but their claimed ethnicity is currently Evenk. This an interesting case of ethnic identification caused by immigration and intermarriage.

11 Koryak and Chukchi

Two major lineages, N1c-M46 and Q-M242, were observed in the Chukchi population. Besides these two lineages, C3*-M217 also comprise a considerable frequency in the Koryak paternal gene pool. Deep sequencing indicated that these C3*-M217 samples in Koryaks belong to C3c2-B90, a rare lineage found in the Evenks and Yakuts. This rare lineage may represent the remains of an ancient population in Northeast Asia.

12 Manchu and Other Tungusic-Speaking Populations

Similar to the situation of the Evenks and Evens in Siberia, C3c-M48 is also the predominant paternal lineage in Tungusic-speaking populations living in the Amur River region (Heilongjiang River in Chinese), including the Oroqen, Hezhen, Nanai, Negidal, Udegey etc. The only exception is Manchu (see Table 1). In Manchu, the frequency of C3c-M48 is low and there is high frequency of haplogroup O-M175. This is consistent with the intense admixture between the Manchu and Han Chinese ethnicities. An interesting fact was found when we started to study the paternal lineage of Aisin Gioro, the imperial house of the Qing dynasty which was founded by the Manchu ethnicity. The paternal lineage of Aisin Gioro is confirmed to be C3b2b1*-M401 (xF5483) rather than C3c-M48 (Yan et al. 2015). Further sequencing showed that this lineage is the brother branch of C3-Star cluster which was considered to be the clade of Chengis Khan or his close relatives

(unpublished date from our lab). This finding is unexpected, but may be consistent with the legend of the Aisin Gioro clan regarding their origin. As argued by a textual research (Yao and Sun 2012), the origin location of the Aisin Gioro clan is not Baekdu Mountain (Changbai Mountain in Chinese) as they claimed, but the border region of the middle reaches of the Amur River near Heihe City in Heilongjiang Province, China. This city, which used to be called Aihui City, was surrounded by Daur populations from the 14th century to the 17th century. Regarding the genetic data on this connection, we do find higher frequency of potential C3b2b1*-M401 (xF5483) samples in Daur populations based on Y-STR haplotypes from Lihong Zheng et al. (2009). Y-SNP testing of Daur population samples is needed to confirm this STR prediction. Beside the legend mentioned above, there are no official history records on the early history of the Aisin Gioro clan. Hence, according to genetic data, we propose that the Aisin Gioro clan probably changed their language from a Daur-like language to a Tungusic language during the long-distance migration from the border region of the middle reaches of the Amur River to Hetuola City in Liaoning Province, China.

13 Discussion

According to the latest achievements of genetic studies, there are *s* founding paternal lineages for Uralic populations: N2a1-L1419, N2a1a-B169, N3a1-B211, N3a3a-L550, N3a3b-VL39, and N3a4-Z1936. More studies on the expansion times of these lineages and sampling from a vast region are needed to explore the detailed ethnic history of Uralic populations and the founding process of this language family. In North Eurasian populations, there are some other brother branches of these lineages. These lineages represent the demographic background of North Eurasia. Most of them have become important parts of other modern ethnic groups in Siberia. So, they are keys to understanding the history of Turkic and Mongolic-speaking populations and their languages.

Until now, the available genetic data is not detailed enough to determine the founding paternal lineages of Turkic language group populations. Various predominant lineages were observed in different Turkic-speaking populations, such as Q-M25 in Turkmen, C3*-Star cluster in Kazaks, R1a1a-M17 in Kyrgyz and populations in Altai Mountain Region, N1b-P43 and Q-L53 in Tuvan/Tofalar and N1c-M46 in Yakuts. The genetic pattern we have now observed is extremely consistent with the proposed “elite dominant mode” for expansion of Turkic languages. Ancient DNA may help to understand the early history of Turkic-speaking populations and their languages.

For Mongolic-speaking populations, current genetic data shows that there were three founding paternal lineages, including C3*-F1918 (previously known as Star Cluster), C3c-M48, and C3d-M407. Haplogroup C3-F1756, characterized by a deletion on DYS448, is also distributed widely in Mongolic-speaking populations, even though its frequency is low. According to the available data, these four

lineages originated from different regions before they integrated and became a common ancestor group of all present-day Mongolic-speaking populations. More work on these four lineages and ancient DNA tests are needed to understand the detailed history of Mongolic-speaking populations and their languages.

There is little Y-chromosome sequencing data for Koryak and Chukchi populations. These two populations are generally admixtures of ancient populations with predominant lineage Q-M242 and ancient populations with predominant lineage N1c-M46. The N1c-M46 clade in Koryak, Chukchi, and Eskimo populations split from its brother clade at about 4900 years ago and started its expansion about 2700 years ago, as shown in Fig. 3.

In conclusion, the current available genetic evidence from Siberian populations outlines a general scenario of dispersal and expansion patterns of various paternal lineages throughout Siberia. Populations from the same language group tend to have their own predominant paternal lineages. In this research, based on the comparison of Y-chromosome haplogroup frequencies and dating of each sub-clade among different populations, we proposed a series of language replacement events during the founding of Siberian populations and their language family and/or group. However, sequencing more samples from ethnic groups and testing ancient DNA are needed to form a solid conclusion about the complex demographic history of Siberian populations and their languages.

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